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- (71) Applicant: EXACT LABORATORIES, INC. [US/US]; 63 Great Road, Maynard, MA 01754 (US). (88) Date of publication of the international search report: 30 August 2001
- (72) Inventors: SHUBER, Anthony, P.; 11 Grant Street, Milford, MA 01757 (US). PIERCEALL, William; 17 Abbott Street, Wellesley, MA 02482 (US). For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.



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(54) Title: METHODS FOR DETECTING MUTATIONS USING PRIMER EXTENSION

(57) Abstract: Methods for detecting nucleotide deletions in biological samples are described. Methods of the invention are particularly useful for detecting deletions in regions of polynucleotide repeats. In particular, methods of the invention are useful to detect deletions at the BAT26 locus.

## INTERNATIONAL SEARCH REPORT

Int .tional Application No

PCT/US 00/21763

A. CLASSIFICATION OF SUBJECT MATTER  
IPC 7 C1201/68

According to International Patent Classification (IPC) or to both national classification and IPC

## B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)

IPC 7 C12Q

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Electronic data base consulted during the international search (name of data base and, where practical, search terms used)

EPO-Internal, WPI Data, PAJ, CHEM ABS Data, MEDLINE, BIOSIS, EMBASE

## C. DOCUMENTS CONSIDERED TO BE RELEVANT

| Category * | Citation of document, with indication, where appropriate, of the relevant passages   | Relevant to claim No. |
|------------|--|-----------------------|
| X          | ANDREAS BRAUN ET AL: "Improved Analysis of Microsaellites Using Mass spectrometry" GENOMICS, vol. 46, 1997, pages 18-23, XP002162453 | 1-12, 22-30           |
| Y          | page 19, left-hand column, line 59 -page 19, right-hand column, line 3; figure 2   | 13-21                 |
| Y          | US 5 741 650 A (LAPIDUS STANLEY N ET AL) 21 April 1998 (1998-04-21) cited in the application claims 1-5                              | 13,14                 |

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☒ Further documents are listed in the continuation of box C.☒ Patent family members are listed in annex.

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Date of the actual completion of the international search

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Name and mailing address of the ISA

European Patent Office, P.B. 5818 Patentlaan 2  
NL - 2280 HV Rijswijk  
Tel. (+31-70) 340-2040, Tx. 31 651 epo nl,  
Fax: (+31-70) 340-3016

Authorized officer

Gabriels, J

## INTERNATIONAL SEARCH REPORT

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| C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT |   |                       |
|--|---|-----------------------|
| Category *   | Citation of document, with indication, where appropriate, of the relevant passages  | Relevant to claim No. |
| Y  | HOANG JEAN-MARC ET AL: "BAT-26, an indicator of the replication error phenotype in colorectal cancers and cell lines."<br>CANCER RESEARCH,<br>vol. 57, no. 2, 1997, pages 300-303,<br>XP002162454<br>ISSN: 0008-5472<br>page 300, right-hand column, line 35 -page<br>302, right-hand column, line 21<br>---- | 15-21                 |
| X  | US 5 650 277 A (NAVOT NIR ET AL)<br>22 July 1997 (1997-07-22)<br>figure 1; example 1<br>----  | 1-12,<br>22-30        |
| X  | WO 96 30545 A (APPLIED GENETICS INC)<br>3 October 1996 (1996-10-03)<br>page 12, line 22 -page 47, line 3; claims<br>1-33<br>----  | 1-12,<br>22-30        |
| Y  | GISMONDI VIVIANA ET AL: "Characterization of 19 novel and six recurring APC mutations in Italian adenomatous polyposis patients, using two different mutation detection techniques."<br>HUMAN MUTATION,<br>vol. 9, no. 4, 1997, pages 370-373,<br>XP002162567<br>ISSN: 1059-7794<br>table 2<br>----           | 19                    |
| E  | WO 00 70096 A (EXACT LAB INC)<br>23 November 2000 (2000-11-23)<br>the whole document<br>-----   | 1-30                  |

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# INTERNATIONAL SEARCH REPORT

Information on patent family members

International Application No

PCT/US 00/21763

| Patent document<br>cited in search report |   | Publication<br>date | Patent family<br>member(s) | Publication<br>date |
|---|---|---------------------|----------------------------|---------------------|
| US 5741650                                | A | 21-04-1998          | AU 704696 B                | 29-04-1999          |
|   |   |                     | AU 2240697 A               | 22-08-1997          |
|   |   |                     | CA 2215263 A               | 07-08-1997          |
|   |   |                     | EP 0817968 A               | 14-01-1998          |
|   |   |                     | US 5952178 A               | 14-09-1999          |
|   |   |                     | WO 9728450 A               | 07-08-1997          |
| <hr/>                                     |   |                     |                            |                     |
| US 5650277                                | A | 22-07-1997          | IL 102382 A                | 04-08-1996          |
|   |   |                     | IL 111268 A                | 09-05-1999          |
|   |   |                     | AU 4770393 A               | 31-01-1994          |
|   |   |                     | EP 0648222 A               | 19-04-1995          |
|   |   |                     | FI 946127 A                | 20-02-1995          |
|   |   |                     | IL 106199 A                | 24-09-1998          |
|   |   |                     | IL 111267 A                | 14-07-1999          |
|   |   |                     | JP 8507198 T               | 06-08-1996          |
|   |   |                     | NO 945100 A                | 22-02-1995          |
|   |   |                     | WO 9401447 A               | 20-01-1994          |
|   |   |                     | US 5710028 A               | 20-01-1998          |
| <hr/>                                     |   |                     |                            |                     |
| WO 9630545                                | A | 03-10-1996          | AU 5296496 A               | 16-10-1996          |
|   |   |                     | US 5976798 A               | 02-11-1999          |
| <hr/>                                     |   |                     |                            |                     |
| WO 0070096                                | A | 23-11-2000          | AU 5027400 A               | 05-12-2000          |
| <hr/>                                     |   |                     |                            |                     |

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